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Der Laborbereich ist akkreditiert nach DIN EN ISO 15189.

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FDNA project - **Help Geneticists identify patients with small supernumerary marker chromosomes earlier**

Dear parent or sSMC-carrier,

We are supporting a project in sSMC patients working with a company called FDNA (Facial dysmorphology novel analysis). As you may be aware, many genetic diseases are associated with the gradual development of distinctive facial characteristics in affected patients. FDNA have developed specialized software that analyses facial images with the aim of being able to use this facial image analysis to identify facial morphology associated with rare diseases. The software developed by FDNA converts facial images into a mathematical algorithm that describes the characteristics of each facial feature and the spatial relationship of facial features e.g. width of the bridge of the nose, thickness of the lips, distance between the nose and the corners of the eyes and distance from the tip of the nose to the corners of the mouth. FDNA are collecting data from images of patients with many different genetic diseases and have developed their software to be able to successfully differentiate patients with a number of rare diseases. FDNA do not retain any of the children's or patient's images, the photos will be scanned and the only information retained is the numerical information, so all the data is automatically anonymized.

The FDNA software is currently not able to distinguish patients with sSMC from normal patients and/or sSMC-patients among each other. The aim of this current project is to see whether they can develop the software further to be able to distinguish between patients with different types of sSMC, e.g.

such leading to cat-eye syndrome, i(18p)-syndrome, Pallister-Killian syndrome, i(15q)-syndrome and Emanuel-syndrome. To be able to do this they need to collect data from as many facial images as possible from children at different ages with different types of sSMC.

We invite you to participate in this project and upload photographs of children with sSMC into this secure portal https://community.fdna.com/sSMC
Access code: sSMC!15

Alternatively you may send the photos to me, Thomas.Liehr@med.uni-jena.de and I can upload for you.

Serial photographs of the same child / sSMC carrier at different ages are particularly useful for showing changes with aging process, so if possible please provide e.g. past school photograph type photos at different ages.

Thank you for taking the time to read about this project. There is no obligation for you to take part in this project if you do not wish to.

Sincerely

Patient Informed Consent

I _____ hereby agree to have the digitized photographs, clinical information and family history information of _____ [Name of Patient] to be recorded, processed and stored, using an automatic dysmorphology analysis software accessible securely only by the below mentioned scientist. Face2Gene is a genetic search and reference tool, powered by the Facial Dysmorphology Novel Analysis technology. Face2Gene facilitates detection of facial dysmorphic features and recognizable patterns of human malformations to present comprehensive and up-to-date genetic references with the goal to help identification of known syndromes.

Face2Gene is fully HIPAA (= The Health Insurance Portability and Accountability Act) compliant and any patient information uploaded is accessible only to the below mentioned scientist, unless you also agree to one of the following options

- ☐ Information may be also shared with selected physicians, explicitly authorized by the below mentioned scientist for clinical consultations purposes.

- ☐ Information may be also shared with a group of expert physicians for informational and educational purposes, as well as professional commentary.

- ☐ Information may be also shared with all members of Face2Gene's physician network, as well as third parties for informational and educational purposes, as well as professional commentary and medical research (including scientific publications) purposes.

Other than indicated above, personal health information of patients will not be shared or published and you have the right to request no further usage of these data by contacting the following individual:

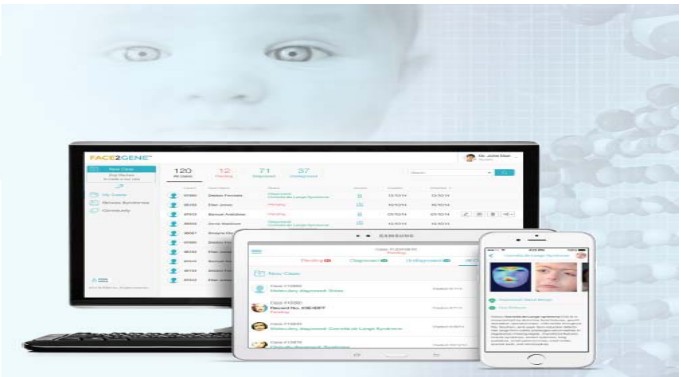
PD Dr. Thomas Liehr, Institute of Human Genetics, Kollegiengasse 10, 07743 Jena, Germany; Thomas.Liehr@med.uni-jena.de

Signature / Guardian's Signature	Date

Relationship (if signed by guardian)

FACE2GENE

FACIAL DYSMORPHOLOGY NOVEL ANALYSIS



WHAT IS THE PURPOSE OF THIS PROJECT?
Many genetic syndromes are associated with the appearance or gradual development of distinctive facial characteristics in affected patients. FDNA has developed a technology that analyzes facial photos to identify facial morphology associated with rare diseases. The Face2Gene computer program converts facial photos into a de-identified mathematical algorithm that describes the characteristics of each facial feature. FDNA is collecting data from images of patients with confirmed diagnosis of many different genetic diseases to improve this technology further. **As more data is collected, the algorithm learns and becomes better, thus enhancing Face2Gene's capabilities for the benefit of the entire genetic expert community and their patients.**

WHAT DO I HAVE TO DO TO HELP?
Parents of children with confirmed diagnosis of genetic syndromes are asked to upload facial photos of their children to this secure and private portal dedicated to help training the technology. Serial photos of the same patient at different ages are particularly useful for showing changes over time, so you are encouraged to provide current and past photos of your child at different ages.
For each photo, please write the estimated age at the time the photo was taken, gender and ethnicity as well as the exact confirmed diagnosis (name of syndrome, specific test results, if available).

WHAT ARE THE POSSIBLE BENEFITS FROM HELPING THIS PROJECT?
The results of this project may improve the scientific understanding of the facial characteristics of multiple cranio-facial syndromes and how these change over time. Results may assist in developing better tools for early diagnostics of multiple genetic syndromes and promote research for drug discovery and effectiveness.

IF I TAKE PART IN THIS PROJECT, HOW WILL MY PRIVACY BE PROTECTED?
No identifying information about patients will be retained by FDNA in this project. All facial photos are converted into de-identified mathematical information. The only information linked is about the syndrome, ethnicity, sex and age of the patient in the respective photo. FDNA applies the highest standards of security (The Health Insurance Portability and Accountability Act = HIPAA) to this online portal to ensure the most stringent privacy compliance.

FDNA Accessible Genetics
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To Learn More and access Face2Gene For Free
Visit: www.Face2Gene.com

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